



## NDRG1 gene

N-myc downstream regulated 1

### Normal Function

The *NDRG1* gene provides instructions for making a protein whose function is not well understood. The NDRG1 protein may play a role in stopping cell growth and prompting cells to mature and take on specialized functions (differentiate). It probably performs these roles by transmitting signals between the nucleus and other parts of the cell. The NDRG1 protein also interacts with other proteins that help regulate the distribution of fats (lipids) in the body.

The *NDRG1* gene is active in cells throughout the body, but its activity is particularly high in specialized cells called Schwann cells. Schwann cells nourish and protect nerve cells, especially the cell extensions (axons) that transmit nerve impulses. Schwann cells also produce myelin, a fatty substance that covers axons and promotes the efficient transmission of nerve impulses. Scientists speculate that the NDRG1 protein is involved in Schwann cell differentiation, myelin maintenance, and the signaling necessary for the survival of nerve axons.

### Health Conditions Related to Genetic Changes

#### Charcot-Marie-Tooth disease

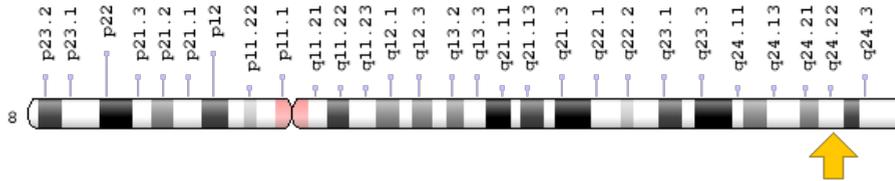
Researchers have identified at least two *NDRG1* gene mutations that cause a form of Charcot-Marie-Tooth disease known as type 4D. One mutation creates a premature stop signal in the instructions for making the NDRG1 protein, which results in an abnormally short protein. The other mutation leads to the production of a protein that is missing a critical segment. Both of these mutations probably result in a nonfunctional protein.

It is unclear how *NDRG1* gene mutations lead to type 4D Charcot-Marie-Tooth disease. Schwann cells use large amounts of lipids and proteins to make myelin, and they rely on a sufficient supply of these substances. *NDRG1* gene mutations may impair the NDRG1 protein's interactions with other proteins that help distribute lipids in the body. As a result, the supply of lipids to Schwann cells may be disrupted, affecting myelin production and altering the transmission of nerve impulses. *NDRG1* gene mutations may also disturb signaling that is necessary for the survival of axons. Loss of myelin and problems with nerve impulse transmission are signs of type 4D Charcot-Marie-Tooth disease.

## Chromosomal Location

Cytogenetic Location: 8q24.22, which is the long (q) arm of chromosome 8 at position 24.22

Molecular Location: base pairs 133,237,171 to 133,297,587 on chromosome 8 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- CAP43
- CMT4D
- differentiation-related gene 1 (nickel-specific induction protein)
- DRG1
- GC4
- HMSNL
- N-myc downstream regulated gene 1
- NDR1
- NDRG1\_HUMAN
- NMSL
- protein regulated by oxygen 1
- PROXY1
- reducing agents and tunicamycin-responsive protein
- RIT42
- RTP
- TARG1
- TDD5

## **Additional Information & Resources**

### Educational Resources

- Basic Neurochemistry (sixth edition, 1999): The Schwann cell is the myelin-producing cell of the peripheral nervous system  
<https://www.ncbi.nlm.nih.gov/books/NBK28217/#A42>

### GeneReviews

- Charcot-Marie-Tooth Neuropathy Type 4  
<https://www.ncbi.nlm.nih.gov/books/NBK1468>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28NDRG1%5BTIAB%5D%29+OR+%28N-myc+downstream+regulated+gene+1%5BTIAB%5D%29%29+OR+%28%28CAP43%5BTIAB%5D%29+OR+%28DRG1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- NMYC DOWNSTREAM-REGULATED GENE 1  
<http://omim.org/entry/605262>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
<http://atlasgeneticsoncology.org/Genes/NDRG1ID41512ch8q24.html>
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=NDRG1%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=7679](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7679)
- Inherited Peripheral Neuropathies Mutation Database  
<http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=7>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/10397>
- UniProt  
<http://www.uniprot.org/uniprot/Q92597>

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